Glossary

allele Any one of the variations of a gene or a polymorphic DNA marker found in the members of a species. Numerous alleles of a given gene or polymorphic DNA marker may exist, but an individual eukaryote possesses at most two alleles of the gene or polymorphic DNA marker.

Alu sequence A DNA sequence about 300 base pairs long that is repeated, one copy at a time, almost 2 million times along the DNA molecules of the human genome. Sequences similar to the Alu sequence are dispersed throughout other mammalian genomes. The function of such repeated sequences is not known.

amino acid An organic compound consisting of a hydrogen atom, an amino group ($-NH_2$), a carboxyl group (–COOH), and a "side chain" bonded to a carbon atom. Each of the more than eighty known amino acids contains a different side chain. Twenty of the known amino acids serve as the building blocks of proteins.

autosome Any chromosome of a eukaryotic organism other than a sex chromosome. The human genome includes forty-four autosomes as twenty-two homologous autosome pairs. Compare **sex chromosome**.

bacteriophage Also called phage. Any one of the viruses that infect bacteria. The genomes of certain phages have been modified to serve as cloning vectors. Fragments of foreign DNA with lengths between about 12,000 base pairs and about 22,000 base pairs are cloned in vectors derived from the genome of the λ (lambda) phage.

base sequence See **sequence**.

biosynthesis Formation of chemical compounds by a living organism.

cDNA (complementary DNA or copy DNA) A single-stranded DNA segment whose sequence is complementary to that of a messenger RNA and thus consists of sequences complementary to the protein-coding sequences that appear on the sense strand of the protein gene from which the messenger RNA was transcribed. cDNAs are synthesized in vitro by reverse transcription of messenger RNAs extracted from cells and are used as hybridization probes for protein genes.

centimorgan The unit of genetic distance, a measure of how frequently two genes on the same chromosome are separated by crossing over and therefore not inherited together. A distance of 1 centimorgan between two genes means that they have a 1 percent chance of not being inherited together. In humans 1 centimorgan corresponds roughly to a physical distance of 1 million base pairs. Compare **genetic distance**.

centromere 1. The DNA sequence within a eukaryotic chromosome to which fibers of the mitotic spindle attach during mitosis and meiosis. Centromeres are essential to the proper parceling out of chromosomes to daughter cells and gametes. 2. The region of attachment between the two identical eukaryotic chromosomes formed by DNA replication. The centromeric region of a chromosome is the constricted region seen along a metaphase chromosome when viewed with an optical microscope; despite the name, the centromeres of many chromosomes are not centrally located. See pages 10–11.

chimera A recombinant clone each member of which contains segments of a genome that are noncontiguous in vivo.

chromatin The complex of DNA and five proteins (histones) that is the major component of eukaryotic chromosomes.

chromosome Any one of a certain species-specific number of threadlike cellular structures, each containing a DNA molecule. Every cell of every member of a eukaryotic species possesses the same number of chromosomes, each of which is confined within the cellular nucleus and contains a linear DNA molecule. The chromosomes within a eukaryotic cell are visible with an optical microscope only when they have become condensed in preparation for cell division. In contrast, every member of any prokaryotic species possesses only one chromosome, which contains a circular rather than a linear DNA molecule, is not confined within a nucleus, and is never visible with an optical microscope.

chromosome banding pattern A pattern of alternating dark and light transverse regions (bands) formed on a fully condensed chromosome by appropriate chemical treatment and staining. Such banding patterns are the means for unambiguously distinguishing the different pairs of homologous chromosomes of an individual eukaryote, and the different bands along a single chromosome are used to identify different regions of the chromosome. See page 11.

classical genetics The study of inheritance based on the concept of the gene as a discrete unit of heredity, data about the transmission of genes from one generation to another, and the behavior of chromosomes rather than on the molecular details of genes and chromosomes. Compare **molecular genetics**.

clone 1. A population of genetically identical unicellular organisms (in this

publication *Escherichia coli* or *Saccharomyces cerevisiae*) or viruses (here phages) arising from successive replications of a single ancestral unicellular organism or virus. 2. A recombinant clone. 3. The fragment of foreign DNA contained in each member of a recombinant clone. 4. A population of identical cells arising from the culture of a single cell of a certain type, such as a human fibroblast or a rodent-human hybrid cell containing a full set of rodent chromosomes and a single human chromosome.

cloning See **molecular cloning**.

cloning vector A relatively short length of DNA into which a DNA fragment to be cloned is inserted. The resulting recombinant vector can be replicated by a host cell by virtue of certain DNA sequences contained within the cloning vector.

codon 1. A triplet of adjacent ribonucleotides along a messenger RNA. Sixty-four such RNA codons are possible. The RNA codon AUG is always the first codon in a messenger RNA to be translated (into the amino acid methionine) and is therefore called the START codon. Each of three other RNA codons signals the cessation of translation of a messenger RNA and is called a STOP codon. Each of the remaining sixty RNA codons is translated into one of the other nineteen amino acids that appear in proteins. 2. The triplet of adjacent deoxyribonucleotides that must appear within the sense strand of a protein gene if transcription of the template strand of the gene is to yield a given RNA codon. The DNA codon corresponding to a given RNA codon is generated simply by replacing each U in the RNA codon by T. See **genetic code**.

complementary base pair Either of the two pairs of bases A and T or C and G. A is said to be the complement of T and vice versa, and C is said to be the complement of G and vice versa. The succession of hydrogen bonds between complementary base pairs is the glue that holds DNA in its natural double-stranded configuration. (Another complementary base pair is the pair A and U, the RNA analogue of T.)

complementary DNA See **cDNA**.

complementary sequences The sequences of two single-stranded nucleic-acid segments (either two single-stranded DNA segments or one single-stranded DNA segment and an RNA segment) are said to be complementary if each base in the 5′-to-3′ sequence of one segment is the complement of the corresponding base in the 3′-to-5′ sequence of the other segment. For example, the DNA sequence 5′-GTAGC-3′ is complementary to the RNA sequence 3′-CAUCG-5′, and the two strands of a chromosomal DNA molecule are complementary along the entireties of their lengths. Two single-stranded nucleic-acid segments with complementary sequences can hydrogen bond to each other.

contig A set of overlapping cloned DNA fragments all members of which have been arranged in the same order that the fragments are found along a chromosomal DNA molecule in vivo. A contig map is a physical map made up of contigs. See pages 118–119.

cosmid A synthetic cloning vector possessing desirable features of plasmid and λ-phage cloning vectors. Fragments of foreign DNA with lengths between about 33,000 base pairs and about 47,000 base pairs can be cloned in cosmids.

crossing over A natural process that produces new combinations of the genetic information present on the DNA molecules within two homologous chromosomes by effecting the exchange, during meiosis, of corresponding regions of the DNA molecules. See page 32.

cytogenetics The study of the inheritance of traits by combining methods of both cytology and genetics. The discovery of microscopically visible chromosomes and advances in optics, microscopy, and cytochemistry have given the field of cytogenetics a prominent place in the study of chromosome number and morphology in both normal and diseased states.

cytology The study of the structure, behavior, reproduction, and pathology of cells.

cytoplasm The substance of a eukaryotic cell between the cellular membrane and the nuclear membrane.

denature To separate DNA into its constituent single strands.

deoxyribonucleotide See **nucleotide**.

diploid chromosome set The species-specific set of chromosomes found in all the cells of a multicellular organism except its gametes. The diploid chromosome set of most species contains an even number of chromosomes because the chromosomes occur as homologous pairs or in the case of those species that possess sex chromosomes, of homologous pairs of autosomes and a pair of homologous or nonhomologous sex chromosomes. One member of each chromosome pair is inherited from each parent. The diploid chromosome set of any human normally contains forty-six chromosomes. Compare **haploid chromosome set**.

DNA (deoxyribonucleic acid) A double-

stranded polymer consisting of a roughly random sequence of four deoxyribonucleotide pairs. Species-specific and individual-specific genetic information is encoded in the order of the deoxyribonucleotide pairs (usually called simply base pairs) along the chromosomal DNA molecule or molecules of an organism. See pages 40–41.

DNA fingerprint Any observable characteristic of a chromosomal DNA molecule or a fragment thereof that can be used to identify a particular individual of a species, a particular recombinant clone, or a particular product of a polymerase chain reaction.

DNA library A collection of recombinant clones. Each member of each recombinant clone in a library contains the same fragment of foreign DNA, as an insert within a recombinant vector, and all the foreign-DNA fragments within all the members of all the recombinant clones in the library originated from the same source.

DNA replication The process by which a parent chromosomal DNA molecule is converted into two daughter DNA molecules, each identical to the parent DNA molecule and each consisting of one strand of the parent DNA molecule and one newly synthesized strand. The two daughter DNA molecules are bound to each other along their centromeric regions and together are called a sister-chromatid pair. See pages 42–43.

DNA sequence A fragment of DNA with a particular, although not necessarily known, sequence.

dominant allele A variant of a gene that determines which variant of the trait specified by the gene is exhibited by an individual eukaryote when only one copy of the allele is present in the genome of the individual. Compare **recessive allele**.

enzyme A protein that acts as a catalyst in a biochemical reaction. A few biochemical reactions are now known to be catalyzed by RNA molecules rather than by proteins. Such catalytic RNA molecules are called ribozymes.

eukaryote Any species or any individual of a species that is a member of the taxonomic kingdom Protoctista, Fungi, Animalia, or Plantae. A eukaryote, whether unicellular or multicellular, is characterized by the presence within the cytoplasm of numerous specialized organelles (see page 8), by the existence of a membrane-bounded nucleus enclosing genetic material organized into multiple chromosomes, and by an elaborate mechanism of cell division involving a mitotic spindle (see page 14–15). In addition, sexual reproduction, a mechanism for increasing the genetic diversity of a species, is common among eukaryotes. Compare **prokaryote**.

exon A protein-coding region of a gene, that is, a base sequence that is translated according to the genetic code into an amino-acid sequence of the gene's protein product. Most protein-coding genes in eukaryotes consist of a series of exons interrupted by introns. Compare **intron**.

flow cytometry A method for sorting cells according to the amount and/or chemical composition of their constituent DNA. The method has been adapted to sorting metaphase chromosome. See page 237.

gamete An ovum or a sperm. Gametes are produced by meiosis of special cells of multicellular organisms and contain a haploid set of chromosomes.

gel electrophoresis A method for separating fragments of DNA or RNA by length. The method involves migration of the DNA fragments through a gel (a porous, semisolid medium) under the influence of an electric field. See pages 55–56.

gene A segment of DNA that contains the information necessary for the controlled biosynthesis of some "gene product." A protein gene contains the infomation necessary for the biosynthesis, by transcription and translation, of a protein or one of the constituent polypeptides of a protein. An RNA gene contains the information necessary for the biosynthesis, by transcription, of an RNA molecule other than the RNA molecules that are translated into proteins or constituent polypeptides. Any individual eukaryote possesses two copies of almost every gene. The two copies may be identical, or they may differ sufficiently to cause some observable difference in the characteristics of the individual. In either case one copy of the gene is located at some position along the DNA molecule within one member of a homologous chromosome pair; the other copy is located at the same position along the DNA molecule within the other member of the homologous chromosome pair. Genes are parceled out to daughter cells and to gametes as predicted on the basis of the observed behavior of chromosomes during mitosis and meiosis.

gene expression Conversion of the information in a gene to a gene product (see **gene**). The rate at which a gene is expressed varies in response to external stimuli and, in the case of multicellular organisms, with cell type and developmental stage.

gene regulation Control of the rate at which a gene is expressed. The primary

mechanism of gene regulation is control of transcription initiation. See page 64.

genetic code A listing of the amino acid or translation signal specified by each of the sixty-four possible codons. See page 48.

genetic distance A quantity that can be roughly correlated with the physical distance between two genes that are located on the same chromosome. The genetic distance between two such linked genes is defined as the probability of the occurrence, during a single meiosis, of crossing over at any point along the segment of DNA between the two genes. The method used to determine a genetic distance, called linkage analysis, requires the existence of at least two alleles for each of the two genes and is applicable not only to linked variable genes but also to linked polymorphic DNA markers. The unit of genetic distance is the centimorgan. See pages 34–35 and pages 86–99.

genetic-linkage map A map showing the genetic distances between pairs of linked variable genes or polymorphic DNA markers.

genetics See **classical genetics**, **molecular genetics**.

genome The totality of the DNA contained within the single chromosome of a bacterial species (or an individual bacterium) or within the diploid chromosome set of a eukaryotic species (or an individual eukaryote). The human genome, for example, consists of approximately 6 billion base pairs of DNA distributed among forty-six chromosomes. Sometimes the term "the human genome" is used to refer instead to the approximately 3 billion base pairs of DNA within the twenty-two different human autosomes and the human X and Y chromosomes. The term "genome" is also applied to the genetic material of a virus, which may be either DNA or RNA.

genotype The pair of alleles of a variable gene possessed by an individual, or the pairs of alleles of any number of variable genes possessed by an individual. The genotype of an individual is a primary determinant of the individual's phenotype.

GT sequence The tandem repeat $(5'\text{-GT})_n$, where n is variable and ranges from 15 to 30. The GT sequence is repeated about 100,000 times throughout the human genome; its function is not known.

haploid chromosome set The sequence-specific set of chromosomes found in all the gametes produced by a multicellular organism and consisting of one (randomly selected) member of each homologous chromosome pair possessed by a eukaryotic species and, if both males and females of the species possess a pair of sex chromosomes, of one (randomly selected) sex chromosome. Thus the haploid chromosome number of most organisms is one-half of its diploid chromosome number. A human gamete normally contains twenty-three chromosomes. Compare **diploid chromosome set**.

heterozygous Possessing one copy of each of two different alleles of a gene or a polymorphic DNA marker. Compare **homozygous**.

homologous chromosomes Chromosomes that, during metaphase, are indistinguishable in size, location of centromere, and banding pattern. The homology of a pair of metaphase chromosomes is due to a very high degree of similarity in the order of the deoxyribonucleotide pairs along their constituent DNA molecules. See pages 10–11.

homologous sequences Two molecules or segments of DNA (or RNA) are said to have homologous sequences if a high percentage of the corresponding base pairs (or bases) in their sequences are identical. For example, many genes of *Mus musculus* and *Homo sapiens* have homologous sequences.

homozygous Possessing two copies of the same allele of a gene or a polymorphic DNA marker. Compare **heterozygous**.

hybridization Hydrogen bonding of two single-stranded segments of DNA with complementary sequences or of one single-stranded segment of DNA and one segment of RNA with complementary sequences. Hybridization is the basis of a technique for identifying, among many different DNA fragments, those fragments that contain a DNA segment of interest. The technique requires the availability of a probe for the segment of interest. See pages 61–63.

in-situ hybridization Hybridization between a free segment of DNA (or RNA) and an intact chromosomal DNA molecule. In-situ hybridization is the basis of a technique for determining the location of a fragment of DNA along an intact chromosomal DNA molecule or the location of an intact chromosomal DNA molecule within the cellular nucleus. See pages 61–63.

interphase The entirety of the eukaryotic cell cycle except the mitotic phase. Because the chromosomes within a cell are not condensed during interphase, the genes along the chromosomal DNA molecules are accessible to transcription. Therefore, most of the biosynthetic activities of a cell, including DNA

replication, occur during interphase. The transition from interphase to the mitotic phase is signaled by the condensation of duplicated chromosomes into microscopically visible structures. See page 9.

intron A region of a protein gene that separates one exon of the gene from another. The introns of a protein gene are transcribed but are excised from the RNA transcript before it is translated. Very few prokaryotic protein genes contain introns, whereas many eukaryotic protein genes contain at least one intron. Introns also occur in genes coding for ribosomal RNAs and some transfer RNAs. See page 64. Compare **exon**.

karyotype A display of the set of chromosomes extracted from a eukaryotic somatic cell arrested at metaphase. The chromosomes are banded and photographed through an optical microscope, and the micrographs of individual chromosomes are arranged by hand into a standard array of homologous chromosomes and sex chromosomes (provided the cell originated from a species possessing sex chromosomes). A karyotype is helpful in revealing chromosomal abnormalities that are symptomatic of various disorders. See page 11.

linked genes Two or more genes that reside on the same chromosome of a eukaryotic organism. The classical method for determining whether two genes are linked requires that both genes have dominant and recessive alleles and involves detecting statistically significant deviations from Mendel's law of independent assortment for the coinheritance of the trait variants specified by the alleles of the two genes. See pages 86–93.

locus The position on a chromosome or on its constituent DNA molecule of a gene or other DNA landmark.

map See **genetic-linkage map** and **physical map**.

meiosis The type of cell division undergone by the precursors of gametes. Meiosis involves two successive divisions of nuclear matter, and each gamete produced possesses a haploid chromosome set. See page 15.

messenger RNA (mRNA) An RNA molecule formed by transcription of the template strand of a protein gene and removal from the resulting primary transcript of any introns present. A messenger RNA serves as a template for translation. See pages 45–47.

metaphase The stage of mitosis or meiosis in which the fully condensed duplicated chromosomes (sister-chromatid pairs) are aligned along the equatorial plane of the dividing cell. See pages 14–15.

metaphase chromosome 1. A fully condensed duplicated chromosome (sister-chromatid pair), or, in other words, a sister-chromatid pair in the configuration it assumes during metaphase of mitosis or meiosis. 2. Either one of the fully condensed sister chromatids in a metaphase chromosome. See pages 14–15.

mitotic spindle The apparatus that directs the motion of chromosomes during mitosis and meiosis.

mitosis The type of cell division by which a unicellular eukaryote reproduces asexually or by which a multicellular organism increases in size and replaces dead cells. The daughter cells produced by mitosis are genetically identical to the mother cell. See page 14.

molecular cloning The production of many identical copies of a DNA fragment by inserting the fragment into a cloning vector and propagating the resulting recombinant vector in a host cell.

molecular genetics The study of the molecular details of the regulated flow of genetic information among DNA, RNA, and proteins and from generation to generation. Compare **classical genetics**.

mutagen Any of a wide variety of agents, including certain types of radiation, certain chemicals, and infecting viruses, that can cause mutations in the genome of an organism.

mutation Any alteration in the base sequence(s) of the constituent DNA molecule(s) of the genome of an organism. Some mutations are caused by external mutagens; others are caused by natural mechanisms such as crossing over or the incorporation of foreign DNA into a genomic DNA molecule. Any nonlethal mutation is transmitted to successive generations of a cell, but only mutations in the DNA of gametes or their precursors are transmitted to successive generations of a multicellular organism.

nematode Any member of a group of worms with unsegmented bodies. The nematode *Caenorhabditis elegans* is the only multicellular organism for which the lineage of every cell has been traced and for which the interconnections among all the neurons have been determined. Furthermore, a physical map covering 95 percent of the *C. elegans* genome has been constructed, and a project to sequence its entire genome is under way.

nucleic acid The generic name for DNA or RNA.

nucleotide The generic name for the building blocks of DNA and RNA. A deoxyribonucleotide (or DNA nucleotide) consists of a phosphate group attached to a deoxyribose residue, which in turn is attached to the residue of one of four nitrogenous organic bases (adenine, cytosine, guanine, or thymine). A ribonucleotide (or RNA nucleotide) consists of a phosphate group attached to a ribose residue, which in turn is attached to the residue of one of four nitrogenous organic bases (adenine, cytosine, guanine, or uracil). Neighboring nucleotides along a strand of DNA or RNA are linked by a covalent bond between an oxygen atom in the phosphate group of one nucleotide and the 3′ carbon atom in the sugar residue of the other nucleotide. See pages 40–41.

oligonucleotide A segment of single-stranded DNA synthesized in vitro and usually containing at most a few tens of deoxyribonucleotides. Oligonucleotides serve as hybridization probes and as the primers required in a polymerase chain reaction.

phage See **bacteriophage**.

phenotype The variant of a heritable trait exhibited by an individual, or the variants of any number of inheritable traits exhibited by an individual. The genotype of an individual cannot in general be deduced from its phenotype. Compare **genotype**.

physical map A map showing physical distances (in base pairs) between landmarks along a chromosomal DNA molecule, such as genes, restriction sites, RFLPs, and sequence-tagged sites. The lowest-resolution physical map of a human chromosomal DNA molecule is a chromosomal banding pattern; the highest-resolution map, the sequence of deoxyribonucleotide pairs along its entire length, will probably not be available for some time. The physical maps to be produced by the Human Genome Project are contig maps (see **contig**), which are of intermediate resolution.

plasmid Small, circular DNA molecules found in and replicated by various bacterial species, including *E. coli*. Engineered plasmids are used as cloning vectors. Fragments of foreign DNA about 4000 base pairs long can be cloned in plasmids.

polymerase An enzyme that catalyses the template-directed linking (polymerization) of the precursors of deoxyribonucleotides or ribonucleotides. A DNA polymerase catalyzes the basic chemical reaction of DNA replication: the synthesis of a strand of DNA with a sequence complementary to that of a template strand of DNA. The reaction requires the pre-existence of a "primer," a very short strand of DNA or RNA bound by complementary base pairing to the template strand. See page 43. An RNA polymerase catalyzes the basic chemical reaction of transcription: the synthesis of an RNA molecule with a sequence complementary to that of one strand (the template strand) of an RNA gene or a protein gene. No primer is required. See page 46.

polymerase chain reaction (PCR) An in-vitro process for producing many millions of copies of a DNA fragment. The process involves successive repetitions of a series of reactions and, when applied to a sample containing many different DNA fragments, can amplify one selected fragment. See pages 128–130.

polymorphic DNA marker A region along a chromosomal DNA molecule of a eukaryotic species whose sequence varies among a population of the species. The alleles of a polymorphic DNA marker are inherited in just the same fashion as are the alleles of a variable gene. Analysis of the co-inheritance of a variable gene on some chromosomal DNA molecule and polymorphic DNA markers on the same chromosomal DNA molecule helps to pinpoint the location of the gene. See pages 94–99.

polypeptide Also called polypeptide chain. A string of amino acids linked by peptide bonds. The term "polypeptide" is not exactly synonymous with the term "protein" because some proteins are composed of more than one polypeptide.

probe A labeled stretch of single-stranded DNA (or RNA) whose sequence includes the complement of one strand of a DNA sequence of interest. Such a probe is required for using hybridization to detect the presence of the DNA sequence of interest in a sample containing many different DNA sequences. See pages 61–63.

prokaryote Any species or any individual of a species that is a member of the taxonomic subkingdom Eubacteria of the kingdom Prokaryotae. Prokaryotes are almost invariably unicellular and are characterized by the absence of a membrane-bounded nucleus, the absence of organelles other than ribosomes, a genome consisting of a single closed loop of DNA, and a mechanism of cell division that does not involve a mitotic spindle. Furthermore, mechanisms for exchange of genetic information among members of a prokaryotic species are rare. Compare **eukaryote**.

promoter The portion of a gene to which RNA polymerase must bind

before transcription of the gene can begin.

protein A biological macromolecule composed of at least one polypeptide. The numerous different proteins specified by the genome of an organism play different roles in its maintenance and reproduction and, in the case of a multicellular organism, in its development from a single cell. Some proteins catalyze the chemical reactions that occur in cells; others provide mobility or mechanical support; others defend against foreign substances; others generate and transmit nerve impulses; and others control cell division and differentiation.

recessive allele A variant of a gene that determines which variant of the trait determined by the gene an individual exhibits only when it is present on both members of a homologous chromosome pair. Compare **dominant allele**.

recombinant clone A clone of a recombinant cloning vector, or, in other words, a clone of a vector than contains a fragment of foreign DNA.

recombinant DNA molecule A stretch of DNA than includes DNA from more than one source and can be replicated by a host cell without being incorporated into the genome of the host cell. Examples are recombinant plasmids and recombinant phage genomes.

repetitive DNA A collective term for all the DNA sequences that occur more than once in the genome of an organism. Prokaryotes possess no or very little repetitive DNA, whereas many eukaryotes possess a great deal. Roughly a third of the human genome, for example, is repetitive DNA. The functions of a few repeated human DNA sequences are known; the functions of most are still a matter of speculation. See **Alu sequence, GT sequence, satellite DNA**.

restriction enzyme (type II restriction endonuclease) A protein capable of binding to any occurrence of a specific short DNA sequence and of catalyzing the cleavage of both DNA strands within or near that sequence. The discovery of restriction enzymes helped precipitate the recombinant-DNA revolution. See pages 52–54.

restriction fragment Any DNA fragment produced by the action of a restriction enzyme.

restriction site Any occurrence of the DNA sequence to which a restriction enzyme binds.

reverse transcription The synthesis of a strand of DNA from a template strand of RNA. The sequence of the synthesized DNA strand is complementary to the sequence of the RNA template strand. Reverse transcription, which is catalyzed by the enzyme reverse transcriptase, is the first step in the reproduction of certain viruses, including the virus that causes AIDS, and is also the reaction by which cDNAs are synthesized in vitro.

RFLP (restriction-fragment-length polymorphism) A type of polymorphic DNA marker that results in differences among individuals in the lengths of the restriction fragments that originate from the polymorphic region.

ribosomal RNA (rRNA) The RNA molecules found in ribosomes.

ribosome A cellular organelle involved in translation. A ribosome effects the synthesis of a protein or a polypeptide by catalyzing the linkage, in the order specified by a messenger RNA, of the amino acids carried by transfer RNA molecules. A ribosome contains a relatively small number of different RNA molecules and a large number of different proteins. See page 47.

RNA (ribonucleic acid) A single-stranded polymer consisting of a sequence of linked ribonucleotides (see **nucleotide**). Numerous different RNA molecules constitute the bulk of the cellular nucleic acid and play different roles. Messenger RNAs, ribosomal RNAs, and transfer RNAs are involved in protein synthesis; others are components of the spliceosomes involved in RNA splicing; a few are known to act as catalysts; and one is known to be involved in the transport of newly synthesized proteins to their ultimate destinations within the cell.

RNA splicing The process by which an RNA molecule transcribed from the template strand of a gene is rid of any introns it may contain. The product of splicing is a messenger RNA or the mature form of some other type of RNA. See page 45.

satellite DNA Any of the tandem repeats found at the centromeric and telomeric regions of the chromosomal DNA molecules of many eukaryotes.

sequence Also called base sequence. 1. A listing of the deoxyribonucleotide pairs within a DNA molecule or segment in the order they appear along the DNA molecule or segment in vivo. 2. A listing of the ribonucleotides within an RNA molecule or segment in the order they appear along the RNA molecule or segment in vivo. The process by which such sequence data are obtained is called sequencing. See pages 151–159.

sex chromosome Males of some species, including all mammals, possess two

chromosomes that are not homologous to each other or to any other chromosomes the males possess. However, one of those exceptional chromosomes is homologous to both members of a homologous chromosome pair possessed by the females of such a species. Any one of those three homologous chromosomes is called an X chromosome. The other exceptional male chromosome is called a Y chromosome. Collectively the X and Y chromosomes are called sex chromosomes because one or the other is involved in determining maleness (possession of testes). The Y chromosome of a mammal is easily distinguished from its much larger X chromosome and is the sex chromosome that determines maleness. On the other hand, the Y chromosome of the fruit fly *Drosophila melanogaster* is comparable in size to its X chromosome and is not the sex chromosome that determines maleness. Although the X and Y chromosomes of a male are not homologous, the two do pair up during meiosis and one or the other is parceled out to each sperm.

sister-chromatid pair The two identical chromosomal DNA molecules formed by replication of a single chromosomal DNA molecule. The binding of the members of a sister-chromatid pair along their centromeric regions accounts for the X shape of metaphase chromosomes.

somatic cell Any cell of a multicellular organism other than gametes or the precursors of gametes.

Southern hybridization A hybridization technique in which the fragments to be interrogated with a hybridization probe have been length-separated by gel electrophoresis and transferred from the gel to a nylon nitrocellulose filter. The filter containing the length-separated fragments is sometimes called a Southern blot. See page 63.

STS (sequence-tagged site) A short (200 to 300 base-pair long) segment of a chromosomal DNA molecule whose sequence has been determined and is known to be unique because the STS can be selectively amplified by a particular polymerase chain reaction. A set of STSs located on a chromosomal DNA molecule helps to integrate the genetic-linkage and physical maps of the chromosomal DNA molecule. See pages 130–134.

telomere 1. Either terminus of a eukaryotic (and hence linear) chromosome. 2. The DNA sequence that terminates either end of a eukaryotic chromosomal DNA molecule.

transcription The biosynthesis of an RNA molecule from a DNA template strand. The sequence of the synthesized RNA molecule is complementary to the sequence of the DNA template strand. See page 46.

transduction The phage-mediated transport of genetic information from a member of a bacterial strain or species to a member of the same or a different strain or species.

transfer RNA (tRNA) Any one of a group of small RNA molecules that are involved in translating the sequence of codons along a messenger RNA molecule into a sequence of amino acids along a polypeptide. See page 47.

transformation The process by which a plasmid cloning vector enters an *E. coli* host cell or a yeast artificial chromosome enters a yeast host cell.

translation The linking of amino acids carried by transfer RNA molecules in an order specified by the order of the codons along a messenger RNA molecule. The product of translation is a protein or a polypeptide. See page 47.

translocation The transfer of a segment of a chromosome from its usual location to a new location on a homologous chromosome or a nonhomologous chromosome. Translocations are often symptomatic of disease and can be detected as changes in the banding patterns or morphologies of metaphase chromosomes.

virus Any of numerous and varied submicroscopic organisms that are incapable of reproduction outside a host cell. The structure of viruses is remarkably simple: each consists of a genome, which may be either DNA or RNA, and a protein body that not only protects the genome but also facilitates entry of the genome into a host cell. Almost all living organisms are susceptible to attack by viruses.

wild type An individual of a species that exhibits the variants of inheritable traits that are typical of a natural population of the species.

YACs (yeast artificial chromosome) 1. A linear recombinant DNA molecule that is replicated as a yeast chromosome by a yeast host cell because it contains, in addition to a fragment of foreign DNA, a yeast centromere, a yeast origin of replication, and two yeast telomeres, one at each end. Fragments of foreign DNA with lengths up to 1 million base pairs can be cloned as YACs. 2. The vector arms to which the ends of a foreign DNA fragment are ligated to form a YAC.

zygote The single cell formed by fertilization of an ovum by a sperm.

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